(19) World Intellectual Property Organization International Bureau





(43) International Publication Date 28 April 2005 (28.04.2005)

(10) International Publication Number WO 2005/038047 A1

(51) International Patent Classification⁷:

C12Q 1/68

(21) International Application Number:

PCT/AU2004/001434

(22) International Filing Date: 20 October 2004 (20.10.2004)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

60/513,097 2003905747 20 October 2003 (20.10.2003)

20 October 2003 (20.10.2003) AU

- (71) Applicants (for all designated States except US): ST VIN-CENT'S HOSPITAL (SYDNEY) LIMITED [AU/AU]; Victoria Street, Darlinghurst, NSW 2010 (AU). VIC-TOR CHANG CARDIAC RESEARCH INSTITUTE [AU/AU]; 384 Victoria Street, Darlinghurst, NSW 2010 (AU).
- (72) Inventors; and
- (75) Inventors/Applicants (for US only): WARD, Robyn, Lynne [AU/AU]; 20 Moncur Street, Woollahra, NSW 2025 (AU). MARTIN, David, I., K. [US/AU]; 7 Glenview Street, Paddington, NSW 2021 (AU). SUTER, Catherine, Mary [AU/AU]; 94 Marion Street, Enmore, NSW 2042 (AU).

- (74) Agent: BLAKE DAWSON WALDRON PATENT SER-VICES; Level 39, 101 Collins Street, Melbourne, VIC 3000 (AU).
- (81) Designated States (unless otherwise indicated, for every kind of national protection available): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NA, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.
- (84) Designated States (unless otherwise indicated, for every kind of regional protection available): ARIPO (BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PL, PT, RO, SE, SI, SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

Published:

with international search report

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

(54) Title: ASSESSMENT OF DISEASE RISK BY QUANTITATIVE DETERMINATION OF EPIMUTATION IN NORMAL TISSUES

(57) Abstract: An assay for assessing the risk of disease in an individual, wherein said assay comprises the steps of isolating a population of cells from normal tissue of said individual, and quantitatively determining the frequency of epimutation of a particular gene in said population of cells, wherein the epimutation of said gene is associated with said disease and said gene is other than one that is subject to normal parent of origin-specific expression. Preferably, the epimutation is DNA methylation of a tumour suprressor gene such as hMLH1, hMSH2, APC 1A, APC 1B and p16.



